



BIN1 gene

bridging integrator 1

Normal Function

The *BIN1* gene provides instructions for making a protein that is found in tissues throughout the body, where it interacts with a variety of other proteins. The BIN1 protein is thought to be involved in the transportation of materials from the cell surface into the cell (endocytosis) and the self-destruction of cells (apoptosis). The BIN1 protein may act as a tumor suppressor protein, which means it prevents cells from growing and dividing too rapidly or in an uncontrolled way.

Several different versions (isoforms) of the BIN1 protein are produced from the *BIN1* gene. These isoforms vary by size and are active in different tissues. The BIN1 protein isoform that is expressed in muscle cells is thought to be involved in the formation of structures called transverse tubules or T tubules. These structures are found within the membrane of muscle cells, where they play a role in muscle tensing (contraction) and relaxation.

Health Conditions Related to Genetic Changes

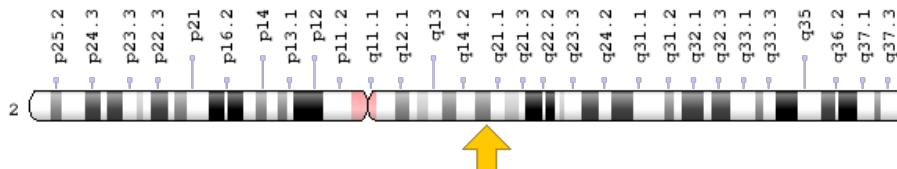
centronuclear myopathy

At least 10 mutations in the *BIN1* gene have been found to cause centronuclear myopathy, a condition that is characterized by muscle weakness (myopathy) in the skeletal muscles, which are the muscles used for movement. Most of these mutations change single protein building blocks (amino acids) in the BIN1 protein. *BIN1* gene mutations result in the production of a protein that cannot form T tubules. A shortage of T tubules in muscle fibers alters their structure, which prevents them from contracting and relaxing normally. The abnormal muscle fibers underlie the muscle weakness characteristic of centronuclear myopathy.

Chromosomal Location

Cytogenetic Location: 2q14.3, which is the long (q) arm of chromosome 2 at position 14.3

Molecular Location: base pairs 127,048,023 to 127,107,400 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AMPH2
- amphiphysin II
- amphiphysin-like protein
- AMPHL
- BIN1_HUMAN
- box-dependent myc-interacting protein 1
- myc box-dependent-interacting protein 1
- SH3P9

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): T tubules and the sarcoplasmic reticulum
<https://www.ncbi.nlm.nih.gov/books/NBK26888/?rendertype=figure&id=A3072>
- Washington University, St. Louis: Neuromuscular Disease Center: Centronuclear Myopathy, Autosomal Recessive
<http://neuromuscular.wustl.edu/syncm.html#arcnm>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28BIN1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- BRIDGING INTEGRATOR 1
<http://omim.org/entry/601248>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
<http://atlasgeneticsoncology.org/Genes/BIN1ID794ch2q14.html>
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=BIN1%5Bgene%5D>
- HGNC Gene Family: N-BAR domain containing
<http://www.genenames.org/cgi-bin/genefamilies/set/1289>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1052
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/274>
- UniProt
<http://www.uniprot.org/uniprot/O00499>

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